

NEWBORN SCREENING AND INFANT HEARING WINTER 2015

VOLUME 1

ISSUE 6



LAB NOTES: BY LESLIE HIMSTEDT

The Newborn Screening Lab is now open on Saturday and we accept samples from 8– 10 AM.

The building is locked on the weekends so instruct your delivery service to call the number posted and someone from the lab will meet the driver at the door to receive the samples.

Sample unsatisfactory rate has significantly increased the last quarter. The primary causes for sample rejection are:

1. QNS– quantity of blood in circle is not sufficient for testing. This is usually due to the blood not soaking completely through to the other side of the filter paper.
2. Circles overfilled or layered– there is too much blood on the filter paper, either outside the circle or the sample is puckered and crusty from too much blood.
3. Filter paper expired– each card is printed with an expiration date. Samples collected on out of date cards cannot be tested. Check the date before you fill out the card and perform the heel stick.

Leslie

SCREENED CONDITIONS FOR WHICH TIME IS CRITICAL & WHICH REQUIRE EMERGENCY ACTION

Endocrine Disorders

Congenital adrenal hyperplasia (CAH): Inc 17-OHP

Other Genetic Disorders

Galactosemia (GALT +) (s): Dec. RBC GALT

Amino Acid Disorders

Maple syrup urine disease (MSUD): Inc. leucine & isoleucine

Citrullinemia (CIT) *Urea Cycle defect/ASA deficiency*: Inc. citrulline

Argininosuccinate lyase deficiency (ASL) or ARG-1: Inc. citrulline
Urea Cycle Defect

Fatty Acid Oxidation Disorders

Medium chain acyl CoA dehydrogenase deficiency (MCAD): Inc. C8, early hypoglycemia esp breast fed, episodic, give carnitine

Very long chain acyl CoA dehydrogenase deficiency (VLCAD): Inc. C14:1

Long chain hydroxyacyl CoA dehydrogenase deficiency (LCHAD or Trifunctional protein deficiency (TFP)): Inc., C16-OH

Carnitine/acylcarnitine translocase (CAT): Inc. C18

Carnitine pantoic transferase (CPT 2): Inc. C18

Organic Acidemias

Propionic acidemia (PROP): Inc. C3

Methylmalonic acidemia (MMA) due to mutase deficiency (MUT): Inc. C3

Methylmalonic acidemia (MMA) due to cobalamin A,B defect (Cbl A,B) : Inc. C3

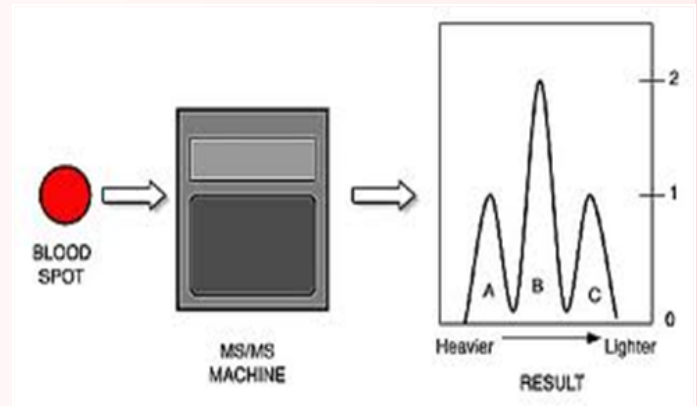
Maternal vitamin B12 deficiency: Inc C3

Isovaleric acidemia (IVA): Inc. C5; “sweaty feet”

Multiple carboxylase deficiency (MCD): Inc. C5-OH

Hydroxy-methyl glutaric acidemia (HMG): Inc. C5-OH

(Glutaric aciduria type 2: numerous Inc. C4 to C12– Not on AR list of disorders screened)



Continued from page 2.....

It is impossible to exaggerate the importance of time in obtaining the results of the dried blood spot newborn screening and taking action.

Please see my PedsPlace presentation given on January 8 entitled “Newborn Screening Emergencies”. This can be viewed at:

<http://media.archildrens.org/Mediasite/Play/4288b5bccfb04d52ac6cdd9bca4f5b721d?catalog=b0fc7897-1802-4ca2-a5ec-186cf3d18d05>

About 10% of the screens which become confirmed disorders each year in Arkansas are the specific disorders which can present clinically as early as 5 days of age. See Screened Conditions for which Time is Critical & which Require Emergency Action). Our goal for these disorders is to have actionable results to you between 5 to 7 days. Outcomes of treatment for these disorders are directly related to the rapidity with which treatment is initiated. Delays in time of drawing the specimen at the hospital, delays in transport of the specimen to the lab, delays in doing the test in the lab, delays in getting the results to you are cumulative.

We are working closely with the birthing hospitals in Arkansas to improve the timeliness of our newborn screening program. You can help by recognizing the urgency of contacting the parent of babies who screen positive for the disorders on this list and checking on the baby’s clinical condition. You will get specific instructions faxed to you from our newborn screening nurses. We all need to work together to prevent deadly delays!

Alan Mease, MD

Medical Director

Newborn Screening Program

“Newborn Screening-focusing on the  of the matter!”

CCHD (Critical Congenital Heart Disease) testing is now one of the tests Arkansas hospitals have implemented! Monday, Jan. 5, 2015 was the start date for recording the Pulse Oximetry Screening Results for Critical Congenital Heart Disease in ERAVE.



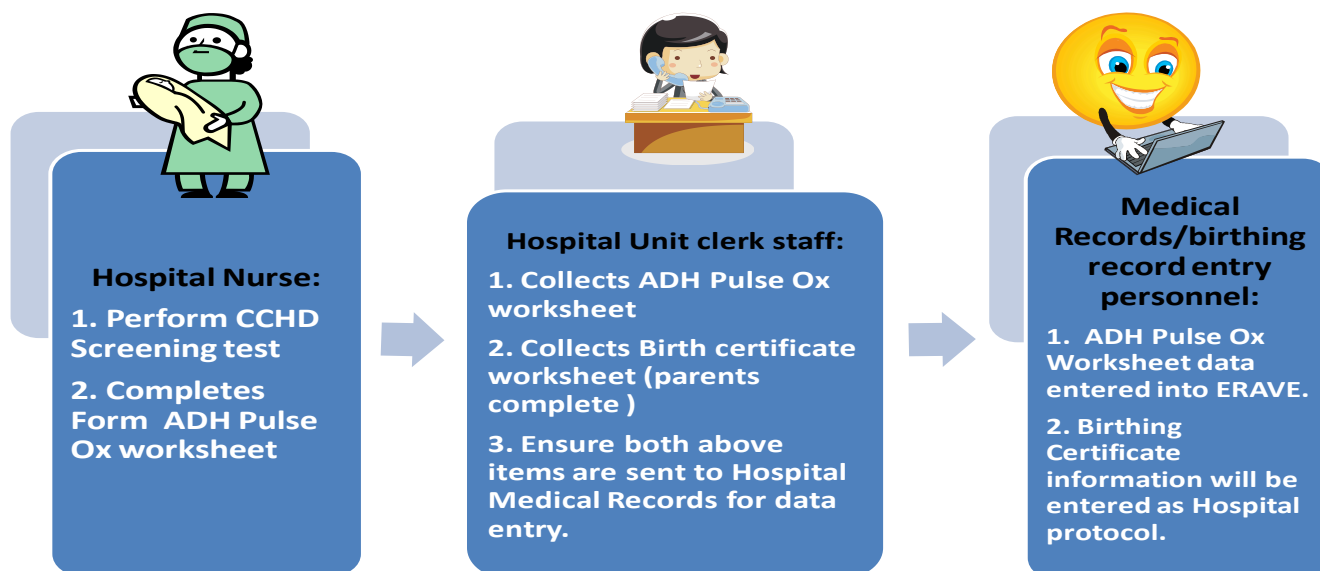
Hospital Birthing Clerks: A big “THANK YOU” to Patsy Bennewise, ADH/ERAVE program Specialist for providing several Webinars/In-service for the hospital birthing clerks! Her contact information: (501)280-4765 or patsy.bennewise@arkansas.gov

Nursery Nurses: We mailed all birthing hospital Administrators & Nursery Managers a packet of CCHD information.

www.archildrens.org/CCHD This web site provides educational tools for health professionals and parents about protocol & procedure.

Below is a “FLOW CHART” suggesting the process if you have any questions e-mail me at: patricia.purifoy@arkansas.gov or call me at: (501) 280-4145.

Recommended Hospital Procedure for AR Dept. Health/Pulse Ox Screening Results Worksheet (CCHD) entry in ERAVE!



FOOD FOR THOUGHT!!!!

“IF YOU WANT TO LIFT YOURSELF UP, LIFT UP SOMEONE ELSE” -Booker T. Washington

ATTENTION:
INFANT HEARING ANNUAL CONFERENCE 2015
APRIL 30TH FROM 9:30 A.M. TO 2:30 P.M.
5800 WEST 10TH STREET, SUITE 902
LITTLE ROCK, AR 72205



INFANT BEING SCREENED BY AUTOMATED
ACOUSTIC BRAINSTEM RESPONSE

**Please Welcome to the Infant
Hearing Program!!!!!!**

Kimberly McCray, IHP Manager



Kim, comes to Child and Adolescent Health after working as a public health educator with ADH Hometown Health Improvement, serving several counties in Northeast Arkansas to help improve awareness and encourage prevention, for the past six years. Originally from Conway, AR, she is a graduate of Arkansas State University- Jonesboro with a B.S. in Exercise Science and a MSHS in Health Science Education. Kim enjoys participating in and watching sports, reading and cooking.

**HOSTED BY DEPARTMENT OF HEALTH THIS
CONFERENCE IS:**

- **OPEN TO ALL HOSPITAL EMPLOYEES WHO UTILIZE HEARING SCREENING EQUIPMENT WITH INFANTS**
- **FREE**
- **6 CEUs AVAILABLE FOR RNs**
- **LUNCH SERVED**
- **REGISTER BY APRIL 1st 2015 @501.280.4740**
- **NEW PRESENTATIONS, INTERACTIVE STATIONS, AABR & OAE EQUIPMENT**
- **COLLABORATING WITH ARKANSAS CHILDREN'S HOSPITAL, UALR, UAMS AND ARKANSAS SCHOOL FOR THE DEAF**



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